ENMC Impact Report 2024

Our year in highlights

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1 Message from Dr Ingeborg Meijer, Chair of the Executive Committee

In 2024, the ENMC returned to full strength after the challenges of the COVID-19 pandemic, resuming in-person workshops to advance the neuromuscular field and benefit those affected by neuromuscular disorders. With renewed inspiration and dedication, nine workshops were successfully hosted in Hoofddorp, the Netherlands. Building on insights gained from the online working period, the workshops now incorporate more comprehensive preparatory work, enhancing their impact and effectiveness.

The workshops covered a variety of neuromuscular disorders, focusing on overarching themes such as guidelines, standards, diagnostics, care, and nomenclature harmonisation. ENMC workshops are not only forums for experts and stakeholders to exchange ideas and discuss research trends but also serve as safe spaces where critical decisions are made by researchers, clinicians, patients, and industry representatives from different countries.

In addition to workshops, the ENMC office attended two key conferences, which provided valuable opportunities to engage with the neuromuscular community, build partnerships, and promote the ENMC's mentoring programme. The first full cycle of the mentoring programme (2021-2024) was successfully completed and evaluated this year, marking an important step in supporting emerging leaders and ensuring long-term capacity in the neuromuscular field.

At the end of 2024, ENMC said goodbye to two esteemed colleagues. After six years as Research Director, Dr Ana Ferreiro stepped down, leaving a legacy of exceptional contributions, leadership, and commitment. She represented ENMC with grace and strength, guiding discussions in the Research and Executive Committees. From January 2025, Prof. Ros Quinlivan (UCL Institute of Neurology, London, UK) will step into this role, and we look forward to her leadership in continuing ENMC's mission.

Another departure was Dr Anna Ambrosini, who stepped down after 20 years as an Executive Committee member representing Fondazione Telethon ETS. Her dedication and steadfast governance were instrumental in keeping ENMC's vision on track. Dr Danila Baldessari will take over this role, bringing fresh energy to the committee.

None of these accomplishments would have been possible without the exceptional work of the Baarn office team - Patricia, Wilma, and Tamara - who ensured smooth operations and secured workshop funding. We also extend heartfelt thanks to the Research and Executive Committees, Associated Partners, workshop organisers and participants, and the Company Forum for their unwavering support and contributions to ENMC's success in 2024.

Dr Ingeborg Meijer,
Chair of the Executive Committee,
Representative of Spierziekten Nederland,
The Netherlands

2 The mission of the ENMC

Over 32 years ago, a group of scientists and clinicians, together with parents of children affected by neuromuscular conditions, launched the European Neuromuscular Centre (ENMC). They had in mind the ultimate goal to improve diagnosis, accelerate the search for effective treatments and improve the quality of life of people with neuromuscular conditions. To achieve this goal, it was, and still is, of utmost importance that experts in this field of (ultra) rare disorders share their knowledge and experience and collaborate in research worldwide.

The ENMC encourages and facilitates this collaborative aim through the organisation of small interactive workshops for multidisciplinary groups of researchers, clinicians and persons affected by neuromuscular conditions, a unique concept in the scientific community.

ENMC Mission Statement

The mission of ENMC is to encourage and facilitate communication and collaboration in the field of neuromuscular research with the aim of improving diagnosis and prognosis, finding effective treatments and optimising standards of care to improve the quality of life of people affected by neuromuscular disorders.

"Connecting people"

3 The ENMC workshops in 2024

In 2024, an almost unprecedented number of 16 workshop applications were submitted to the ENMC. Of these, four were re-applications.

Ten applications were selected for support, of which two workshops took place in 2024 and the remaining eight are planned for 2025.

3.1 Summary of ENMC workshops held in 2024

Workshop no./date and format	Topic	Workshop leaders
Workshop no. 274 19-21 January 2024	ENMC recommendations for optimizing bone strength in neuromuscular disorders	Prof. L. Ward, Prof. A. Moretti, Dr D. Weber, Prof. N. Voermans
Workshop no. 275 9-11 February 2024	Seronegative MG: an update paradigm for diagnosis and management	Prof. A. Evoli, Dr L. Maggi, Prof. J. Palace, Prof. J. Verschuuren
Workshop no. 276 15-17 March 2024	ENMC recommendations on optimal diagnostic pathway and management strategy for patients with exertional rhabdomyolysis worldwide	Prof. P. Laforêt, Prof. J. Vissing, Prof. N. Voermans, Dr S. Bhai
Workshop no. 277 21-23 June 2024	Congenital myopathies: revising and revisiting nomenclature and diagnostic guidelines	Dr J. Dowling, Dr C. Bönnemann, Dr E. Oates, Dr A. Ferreiro
Workshop no. 278 20-21 September 2024	European standards for harmonization of myasthenia gravis (MG) registries and emerging digital solutions	Prof. S. Sacconi, Dr R. Mantegazza, Dr E. Cortés-Vicente, Prof. A. Meisel
Workshop no. 281 4-6 October 2024	2nd ENMC workshop on exercise training in muscle diseases; towards consensus-based recommendations on exercise prescription and outcome measures	Dr E. Voorn, Prof. J. Vissing, Prof. A. Lucia
Workshop no. 279 1-3 November 2024	Classification, clinical care, outcome measures and biomarkers in childhood onset FSHD: towards standardising clinical care and ensuring clinical trial readiness	Dr C. Erasmus, Prof. K. Mathews, Dr K. de Valle, Prof. T. Willis
Workshop no. 282 8-10 November 2024	Standards of diagnosis and care for the sarcoglycanopathies	Prof. J. Díaz-Manera, Prof. E. Pegoraro, Dr T. Stojkovic, Dr L. Lowes
Workshop no. 280 22-24 November 2024	Diagnostic criteria and outcome measures in primary mitochondrial myopathies	Prof. M. Mancuso, Prof. C. Kornblum

Note: The workshop number (no.) is given once an application is officially approved and does not always reflect the order in which workshops are held.

274th ENMC International workshop: ENMC recommendations for optimising bone strength in neuromuscular disorders

Background

In various neuromuscular diseases (NMDs), bone strength may be compromised. There are persistent gaps in knowledge about the development and maintenance of bone strength in patients with NMDs. Enhanced knowledge is needed to better understand multiple aspects such as: the natural history of bone fragility in this context, optimal diagnosis and monitoring strategies, best candidates for treatment, the treatment strategies that provide the most acceptable benefit-risk profile to patients, and the most appropriate outcome measures for gauging treatment responses in the clinical and drug trial settings.

Aims of the Workshop

The goals of the workshop were to summarise the literature, identify knowledge gaps and create plans to improve clinical care and guide future research.

Workshop Deliverables

A survey showed that many people with NMDs suffer from bone fractures and/or osteoporosis and that the prevention and treatment of bone health issues are not part of the usual care provided to people with NMDs. A survey among clinicians showed moderate to good awareness of the importance of bone strength, but lack of medical training on this topic. The current clinical practice appeared to be variable with respect to diagnostics and treatment.

Terminology, definitions, and normal bone strength development were discussed. In clinical practice and research, various methods are used to indirectly estimate bone quality and strength. However, using these tests can be challenging.

For preventive bone-targeted treatment, pharmacological therapies and physical activity have been recommended. However, there is no consensus on when to start and how long to continue bone-protective therapies. As the most widely used agents in clinical practice to prevent and treat bone fragility irrespective of underlying disease, the safety and efficacy of bisphosphonates has been studied for decades. While the use of bisphosphonates in people with NMD has been adapted from observations in other disease settings, their long-term efficacy and safety data specifically in NMD is lacking.

Most of the bone-related evidence in NMDs has been developed in the paediatric setting.

Information on the monitoring and treatment of bone health issues in NMDs is still emerging and is not included in current care recommendations.

In contrast, in the general population, most research and clinical care guidelines have been focused on the prevention and treatment of post-menopausal osteoporosis. There is a knowledge gap on bone strength in adolescence and early adulthood in patients who do not have normal motor abilities.

Educating clinicians and patients on bone issues is crucial to implement bone management in all NMD.

The patient's voice was well covered at this important meeting by Ms Ingrid de Groot, Ms Silke Schlüter and Ms Madelon Kroneman.

275th ENMC International workshop: Seronegative MG: An update paradigm for diagnosis and management

Background

Myasthenia Gravis is an autoimmune disorder caused by autoantibodies targeting antigens of the neuromuscular junction. However, around 10-15% of patients have no detectable autoantibodies, a condition named Seronegative Myasthenia Gravis (SNMG). Clinical recognition and early diagnosis are crucial for the use of appropriate treatments. Establishing the diagnosis in patients with no detectable antibodies may be very challenging, with a substantial risk of misdiagnosis with conditions that mimic several symptoms, such as congenital myasthenic syndromes (CMS) or mitochondrial myopathies. Moreover, detection of antibodies is also relevant for the access to innovative therapies recently approved for the treatment of Myasthenia Gravis.

Organisers of the 275th ENMC International workshop on Seronegative Myasthenia Gravis. From left to right: Prof. Jan Verschuuren (Netherlands), Prof. Jacqueline Palace (UK), Prof. Amelia Evoli (Italy), Dr Lorenzo Maggi (Italy).

Aims of the Workshop

The workshop's main goals were:

1. to provide a diagnostic algorithm for SNMG,
2. to summarise current knowledge about pathophysiology and natural history of SNMG and identify open questions to be addressed in future research studies, and
3. to define patients' unmet clinical needs.

Workshop Deliverables

The workshop started with a series of talks addressing the core topics of the workshop. Thereafter, the talks and the discussion were focused on the differential diagnosis of SNMG, CMS and paediatric MG. During the second day, several aspects of SNMG were covered, including antibody testing techniques, electrodiagnostic studies and other diagnostic tools, demographic and clinical data on SNMG, as well as conventional immunotherapy and novel target treatments. During the last day, the workgroup developed a diagnostic flowchart for SNMG with clear advice on antibody assays as well as other tests, and a focus on the clues for the differential diagnosis of SNMG. The availability of a diagnostic flow chart will likely lead to an improved diagnosis of SNMG and recognition of alternative diagnoses, like e.g. congenital MG or a functional neurological disorder, ultimately resulting in improved patient care. The future projects identified were to collect more data on natural history and treatment effects in SNMG, to create a European study group on SNMG, and to further evaluate and compare the performance of different antibody assays for SNMG serological diagnosis.

276th ENMC International workshop: ENMC recommendations on optimal diagnostic pathway and management strategy for patients with exertional rhabdomyolysis worldwide

Background

Exertional rhabdomyolysis is the uncontrolled breakdown of skeletal muscle due to exercise, which is a potentially life-threatening condition due to the risk of severe kidney failure. Nevertheless, the exact definition of the condition is topic of debate, resulting in very different diagnostic and treatment approaches worldwide. Therefore, guidelines have yet to be harmonised.

Aims of the Workshop

The aim of the workshop was to define an optimal diagnostic pathway and management strategy for patients with exertional rhabdomyolysis.

Workshop Deliverables

Before the workshop, a survey amongst 60 specialists was distributed. The survey results showed that different countries have very different approaches to rhabdomyolysis, so the discussion focussed on formulating very clear criteria for a diagnosis.
The talks focused on new genetic tests and procedures that could help physicians diagnose a possible genetic cause for rhabdomyolysis. It was highlighted that the genes included in the genetic work-up of rhabdomyolysis currently need to be standardised. The role of muscle biopsies in the diagnostic process can be helpful in specific cases, but nowadays genetic testing should be the first approach. Lastly, it was discussed how to support people with rhabdomyolysis after recovery. This is important since there is a risk of developing a second event if individuals start too fast with their daily activities or sports. Managing ongoing symptoms and helping people get back to exercising safely is a topic that requires more attention in clinical practice.

Experts worked together to summarise the gaps in our knowledge about rhabdomyolysis, as a starting point for discussion to develop guidelines on improved ways to diagnose and treat it. In addition, this should ultimately help people with this condition to have better quality of life. It was concluded that the definition of exertional rhabdomyolysis should include symptoms and laboratory tests to identify patients at risk of developing severe complications. At the same time, patients with chronic CK elevation and/or underlying conditions causing recurrent episodes should not be overtreated.

Participants of the 276th ENMC International workshop on rhabdomyolysis.

277th ENMC International workshop: Congenital myopathies: revising and revisiting nomenclature and diagnostic guidelines

Background

Congenital myopathy (CMYO) is the umbrella term used to describe a heterogeneous group of genetic muscle disorders that typically present at birth (even prenatally) or during infancy with hypotonia and muscle weakness. They are typically nonprogressive or only slowly progressive. The initially defining histopathological findings are distinctive structural abnormalities in skeletal muscle fibres without overt dystrophic features. Originally, definitions of CMYOs were based on characteristic muscle biopsy findings. However, given the now evident variability of biopsy findings, the primacy of genetics in current diagnostic approach to this group of disorders, and the fact that many patients with CMYOs may not have had a biopsy, an update/revision of the existing classification and nomenclature of CMYOs is required.

Organisers of the 277th ENMC International workshop on "Congenital myopathies: revising and revisiting nomenclature and diagnostic guidelines". From left to right: Dr Carsten Bönnemann (USA), Dr Ana Ferreiro (France), Dr Emily Oates (Australia), Dr Jim Dowling (Canada).

Aims of the Workshop

The goals of the workshop were to establish an updated nomenclature for congenital myopathies, and to revise the diagnostic guidelines.

Workshop Deliverables

The group began with agreement that the term "Congenital myopathy" should remain the umbrella term, and the acronym CMYO was agreed upon.
A new classification and nomenclature that encompasses different features (gene, mode of inheritance, histotype, unique clinical presentations) is essential, as nomenclature has important mechanistic, treatment-related, and counselling/guidance implications. A precise and accurately named diagnosis is part of each patient's identity and also has implications for the development of therapies, clinical trials, and access to resources. A new framework for nomenclature was formulated by the group. A classification system, distinct from but consistent with the nomenclature, was proposed to include key features as follows:

1. Congenital myopathy;
2. Gene;
3. Inheritance pattern;
4. Main histopathological (or other phenotypical) features (i.e., Congenital myopathy, RYR1-related, AR, with cores).

The diagnosis of CMYO can be made when there are compatible clinical symptoms and either a confirmed genetic diagnosis that accounts for the clinical presentation or muscle biopsy supporting the diagnosis of a congenital myopathy. Moving forward, an updated diagnostic algorithm and supporting guidelines manuscript will be completed by an expert working group.
The meeting concluded by emphasising the need for education and increased awareness of congenital myopathies within the patient and medical communities.

Participants of the 278th ENMC International workshop on Myasthenia Gravis registries.

278th ENMC International workshop: European standards for harmonisation of myasthenia gravis registries and emerging digital solutions

Background

Myasthenia Gravis (MG) is a rare neurological autoimmune disorder that recently gained growing attention due to the discovery of new therapeutic approaches. In order to better assess the value of the new treatments in the context of standard treatments and the ever-improving pathophysiological understanding, large multinational data sets obtained in the real world of MG care are needed. An essential prerequisite for this is the harmonisation of data collection in order to create a solid ground for data exchange and research.

Aims of the Workshop

The main aim of the workshop was to promote a collaboration between patients, clinicians and other stakeholders to identify which tools are needed to build a functional MG registry initiative. The ultimate goal of a MG registry is to improve quality of care for patients, which is achievable through the collection of meaningful large-scale and high-quality data on the European MG population. Moreover, establishing common data elements among different registries is critical for data harmonisation which is essential for collaboration and research initiatives, including epidemiological and clinical studies, clinical trials, pharmacovigilance as well as providing real world data on novel therapies.

Workshop Deliverables

Through a Delphi questionnaire, participants reached a consensus on a list of mandatory and optional data to be included in a MG registry to facilitate data sharing and collaboration.

The importance of a well-structured and comprehensive informed consent form was also taken into account, which must be easy to understand as well as address important issues like data ownership, data anonymity and purpose of data use. The survey showed that in Europe eight MG national registries are already active and two are in the process of being activated, with data of more than 7,000 MG patients being currently available and potentially sharable. Nevertheless, to optimise future collaborations at a European level, several aspects need to be addressed and improved. A further meeting is planned to follow-up on the achieved goals.

279th ENMC International workshop: Classification, clinical care, outcome measures and biomarkers in childhood onset FSHD: towards standardising clinical care and ensuring clinical trial readiness

Background

Facioscapulohumeral muscular dystrophy (FSHD) is a genetic muscle disease. While FSHD is often considered an adult disease, many affected individuals develop symptoms in childhood. Those who present in early childhood often experience more physical limitations and disability. Recent efforts have highlighted the unique needs of children with FSHD, but standards of care and consistent methods of tracking symptoms in this young group have not been established globally. Differences in the way data is collected and symptoms measured make it challenging to compare results across research studies. Clinical trials of potential treatments for adults and older teens with FSHD are underway. It is essential to describe the spectrum of disease in childhood, establish standard guidelines for care, and find accurate ways to measure disease progression to ensure that children can benefit from new treatments as they emerge.

Aims of the Workshop

Workshop aims included describing recognised clinical groups (or phenotypes) and establishing an agreement on classification/terminology and severity markers for children with FSHD, reviewing and refining current care guidelines, and identifying any gaps in knowledge. Additionally, the workshop aimed to pinpoint challenges and facilitators for clinical trials involving children, foster collaboration among healthcare professionals worldwide, and engage with pharmaceutical companies to help shape the design of clinical trials to best serve all children, including those at the more severe end of the spectrum. A survey completed by participants prior to the meeting helped to shape the discussion.

Workshop Deliverables

There was consensus that the term infantile FSHD should no longer be used, that FSHD is a disease spectrum, and it was agreed that when discussing FSHD in children, the population included should be described. It is clear that more research is warranted to establish a paediatric sub-category based on observed and verified metrics. Consensus was also reached on the need for clinical management considerations for paediatric FSHD including (but not limited to) aspects of disease specific clinical care, psychological care, and management of transition. Defining key outcome measures for clinical care, understanding the rate of disease progression, and accounting for the impact of normal growth on outcome measure assessment, and MRI and ultrasound biomarkers requires additional research. Global, standardised protocols and core outcome measures across paediatric FSHD trials are required to allow meaningful comparison of data and ensure consistency across studies.

Organisers of the 279th ENMC International workshop on "Classification, clinical care, outcome measures and biomarkers in childhood onset FSHD: towards standardising clinical care and ensuring clinical trial readiness". From left to right: Prof. Katherine Mathews (USA), Dr Corrie Erasmus (The Netherlands), Dr Katy de Valle (Australia), Prof. Tracey Willis (UK).

280th ENMC International workshop: Diagnostic criteria and outcome measures in primary mitochondrial myopathies

Background

Primary mitochondrial diseases are a diverse and complex group of genetic disorders that affect the structure or function of the mitochondrial oxidative phosphorylation (OXPHOS) system. Primary mitochondrial myopathies (PMM) represent a subset of these diseases that predominantly affect skeletal muscle. These disorders can present at any age, but more severe phenotypes often manifest earlier in life. In 2016, PMM was for the first time clearly defined as a specific entity and recommendations on outcome measures were provided for both children and adults. Numerous clinical trials, based on these outcome measures, have been performed, are ongoing, or are about to start and several observational studies have already been published on PMM since then. In addition, over ten pharmaceutical companies have announced the development of new molecules targeting PMM.

Aims of the workshop

The consensus on PMM needed to be updated and expanded to include new outcome measures, patient-reported outcome measures (PROMs), and new potential biomarkers for natural history studies and interventional clinical trials. In addition, guidance on the use of new digital health technologies in PMM is needed to harness their potential.

Workshop deliverables

The workshop defined PMM and its related phenotypes, evaluated useful outcome measures for natural history assessment (including PROMs), and discussed which of these measures to implement in clinical trials. Gathering more data on the natural history of PMM is crucial for patients and their families as it lays the foundation for future therapies. Moreover, understanding the patient's and family's burden and viewpoint is particularly important, as it can help select appropriate parameters for clinical trials. Including these parameters ensures that trials measure not only the 'academic advances' of investigational medications but also focus on parameters that capture noticeable benefits for patients in their daily lives. This collaborative effort between researchers and patients is essential for meaningful progress in the field of mitochondrial medicine.

Organisers and the Early Career Researchers (ECR) of the 280th ENMC International workshop on Primary Mitochondrial Myopathies. From left to right: Prof. Michelangelo Mancuso (Italy), Luisa Semmler (ECR, Germany), Piervito Lopriore (ECR, Italy) and Prof. Cornelia Kornblum (Germany).

During the workshop weekend, dinner is the time for continuing conversations and fostering new collaborations. Participants of the 280th ENMC International workshop on Primary Mitochondrial Myopathies.

Organisers, Prof. Alejandro Lucia (Spain), Prof. John Vissing (Denmark) and Dr Eric Voorn (The Netherlands) of the 281st ENMC International Workshop on Exercise Training in Muscle Diseases welcomed the participants.

The participants of the 281st ENMC International workshop engaged in some exercise themselves by walking the "Oranje-pad" to support Prinses Beatrix Spierfonds in its mission to take action in the fight against neuromuscular diseases.

281st ENMC International workshop: 2nd ENMC workshop on exercise training in muscle diseases; towards consensus-based recommendations on exercise prescription and outcome measures

Background

Exercise for people with muscle diseases has been investigated increasingly since the last ENMC workshop on exercise in 2008. The beneficial effects and safety of exercise have been established, and exercise is now considered safe and beneficial in the vast majority of muscle diseases. However, patients and health care professionals need guidance on prescribing the frequency, intensity, type, and time of exercise, as well as outcome measures to monitor progress and to evaluate the effects of exercise programmes.

Aims of the workshop

The aim of this workshop was to develop recommendations on exercise prescription and outcome measures. The workshop focussed on aerobic and resistance exercises in muscle diseases. Prior to the workshop, the organisers prepared the outline for a position stand on aerobic and resistance exercise based on the scientific literature. These recommendations formed the centre of discussions in the following areas:

1 exercise prescription,
2 outcomes, and
3 exercise continuation.

Workshop deliverables

In the opening session, the patient representatives presented results of a survey among 2074 patients, and the early career researchers presented results of a survey among 57 healthcare professionals. In the session on outcomes, there was consensus to use exercise testing before an exercise programme to better individualise exercise prescription. Outcomes to monitor progress (including safety) and to evaluate the effects of exercise programmes were discussed. In the last session it was discussed how to keep patients exercising by finding individual motivators for exercise, setting goals, and systematically assessing facilitators and barriers prior to an exercise intervention. This could be facilitated using eHealth technology.

There was consensus that tailored exercise is essentially safe and beneficial for people with muscle disease and should be recommended, including for patients with very weak muscles and for wheelchair users. The recommendations could help patients and health care professionals with planning exercise to improve the health of people with muscle disease.

Participants of the 282nd ENMC International workshop on sarcoglycanopathies.

282nd ENMC International workshop: Standards of diagnosis and care for the sarcoglycanopathies

Background

Limb Girdle Muscular Dystrophy (LGMD) R3 to R6 (sarcoglycanopathies) is a group of four slowly progressive muscular dystrophies. These diseases cause weakness of the muscles around the pelvis and shoulders. They usually start during childhood and worsen over time. However, some patients have a milder disease which starts later and progresses more slowly. Some patients can develop problems with their heart and/or breathing. There are currently no standardised guidelines on how to diagnose and follow up patients in clinic over time. There are also no guidelines about how to care for these patients.

Aims of the workshop

The first objective of the workshop was to develop an algorithm for the diagnosis of sarcoglycanopathies for clinicians, experts, and non-experts in neuromuscular diseases. The second objective was to agree on recommendations for the follow-up, care, and management of the conditions based on published literature and expert opinions.

Workshop deliverables

The group agreed that a confirmed diagnosis can only be established using genetic testing showing two pathogenic variants found in the gene affected. Management of the conditions includes review by a specialist multidisciplinary team of healthcare professionals, including physicians, physiotherapists, occupational therapists, and nutritionists with experience in neuromuscular diseases. Due to the relatively high number of patients who develop heart and breathing complications, specialised doctors in heart and breathing functions should be included in the care of patients as soon as possible. There are no drugs that can slow down the conditions currently. Therefore, the experts recommended physiotherapy, occupational therapy, exercise guidance, nutrition advice, and cardiac and respiratory care as key for the management of patients. The experts also discussed recommendations about use of anaesthesia, emergency care, and the transition from paediatric to adult care. Attention was also paid to the therapeutic interventions for sarcoglycanopathies. Intermittent steroid treatment at a low dosage requires further research to understand its effectiveness.

3.2 Participants at ENMC workshops in 2024

The ENMC strives for a wide range of expertise and experience in its workshop participants to ensure that a broad consensus can be reached at the meetings by having all relevant stakeholders around the table.

For each workshop that took place in 2024, the numbers of different stakeholders are shown in the table below.

No	Workshop Title	Participants	Clinicians	Basic researchers	Translationel researchers	Patients	Patient representatives	Early-Career researchers	Industry	Regulatory	Other*
274	ENMC recommendations for optimizing bone strength in neuromuscular disorders	20	13	1		2	1	3			
275	Seronegative Myasthenia Gravis (MG): an update paradigm for diagnosis and management	28	17		4	1	1	3	2		
276	ENMC recommendations on optimal diagnostic pathway and management strategy for patients with rhabdomyolysis worldwide	25	13	4	4	1	1	2			
277	Congenital myopathies: revising and revisiting nomenclature and diagnostic guidelines	27	13	2	5		4	3			
278	European standards for harmonization of myasthenia gravis (MG) registries and emerging digital solutions	29	15	1	2	2	1	2	4		2
281	2nd ENMC workshop on exercise training in muscle diseases; towards consensus-based recommendations on exercise prescription and outcome measures	22	10	2	4	2	1	3			
279	Classification, clinical care, outcome measures and biomarkers in childhood onset FSHD: towards standardising clinical care and ensuring clinical trial readiness	28	21				3	3		1	
282	Standards of diagnosis and care for the Sarcoglycanopathies	27	15	1	2	2	3	2	2		
280	Diagnostic criteria and outcome measures in primary mitochondrial myopathies	24	18	1	1		1	2		1	
Average of 9 workshops (n)		**26**	**15**	**1**	**2**	**1**	**2**	**3**	**1**	**0**	**0**
%		100%	59%	5%	10%	4%	7%	10%	3%	1%	1%
Total of 9 workshops (n)		**230**	**135**	**12**	**22**	**10**	**16**	**23**	**8**	**2**	**2**

**Other: health economics specialist, health services scientist*

Clinicians, translational researchers and basic researchers formed the majority of the participants (74%), with an average of 59% clinicians, 10% translational researchers and 5% basic researchers per meeting, which reflects the predominantly clinically-orientated nature of the workshops held in 2024.

Connecting basic researchers with clinicians to bridge the lab and the clinic and bring the fundamental science closer to the clinic is one of the aims of the ENMC.

Through the ENMC Patient Participation Programme we aim to ensure that at least 10% of the participants of each workshop are persons affected by a neuromuscular condition, parents or advocates of these patients and/or representatives from a disease-specific patient or funding organisation.

In 2024, these two groups made up 11% of total participants; the patients' voice was well represented. On average, three Early-Career researchers attended each ENMC workshop this year, showing an improvement as compared to previous years. ENMC supports the integration of the next generation of clinicians and basic scientists in established neuromuscular networks via its Early-Career Programme and Mid-Career Mentoring Programme.

In five workshops, where it was relevant, workshop participants included representatives from pharmaceutical companies, the European Medicines Agency, a health economics specialist, and a health services scientist.

New numbers to be proud of in 2024

Patients and patient representatives
This year we welcomed ten patients and sixteen patient representatives (parents, patient associations, patient advocates) to ENMC workshops. They all gave a presentation, asked questions and throughout in the discussions, which was very helpful for the researchers and clinicians in the workshops to learn the patients' needs and interests.

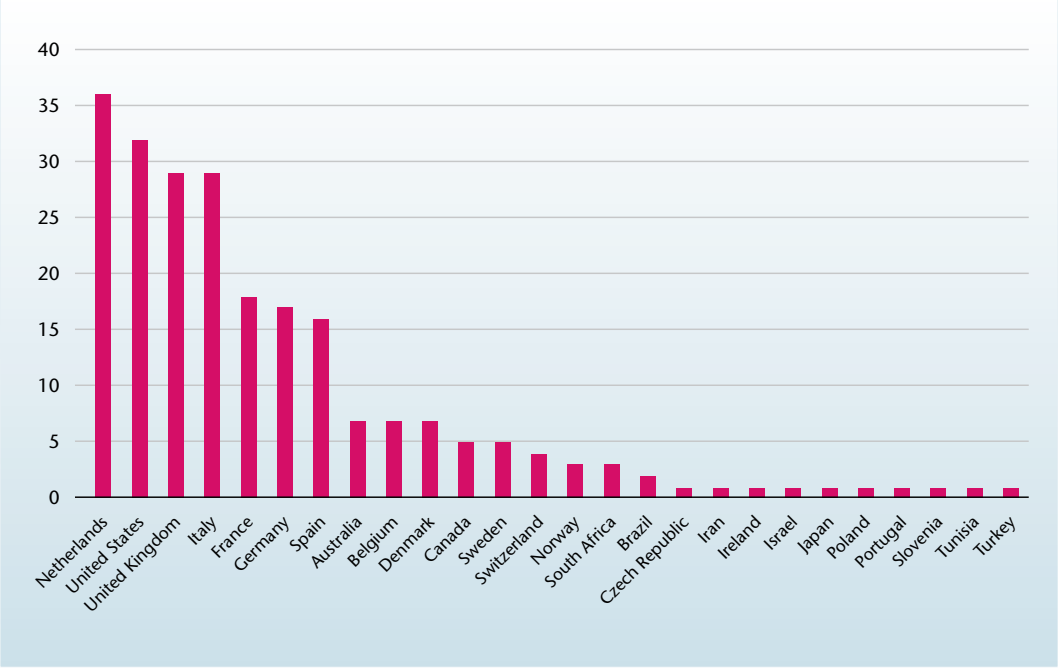
Researchers
We held nine workshops in 2024, with the attendance of 12 basic researchers, 22 translational researchers and 135 clinicians, respectively 5%, 10% and 59% of the total participants. The Early-Career Programme enabled 23 young researchers and clinicians to attend the ENMC workshops and promote their entry in the neuromuscular network.

Sponsors
In 2024, the ENMC was sponsored by eight full partners and five associated partners. The ENMC Company Forum supported our activities through the contributions of ten pharmaceutical companies. We are very thankful for the support from all these partners and acknowledge them with gratitude. In addition to these permanent sponsors, we also would like to thank the great contributions of different organisations that co-sponsored specific workshops in 2024.

3.3 Countries represented in ENMC workshops in 2024

One of the key criteria for a workshop approval by the ENMC is the geographical balance of the participants. The ENMC is convinced that a wide coverage of countries in the workshops is important to make sure that broad consensus is reached at ENMC workshops. This will help to start international research collaborations, ensure the standardisation of healthcare provision for people affected by a neuromuscular condition, and improve the quality of diagnosis and treatment for patients worldwide. In 2024, 26 countries from all over the world were represented at the ENMC workshops (see table below), with a predominance of the ENMC full member countries (The Netherlands, United Kingdom, Italy, France, Germany, Denmark and Switzerland) and some non-member European countries like Spain, Belgium, Sweden and Norway. Non-European countries like USA, Australia, Canada and Brazil were also represented by individual researchers and clinicians at the nine workshops in 2024.

Total number of participants per country in ENMC workshops 2024



4 The ENMC from the perspectives of the outgoing Research Director and a former Executive Committee member

In 2024, we bade farewell to our Research Director, Dr Ana Ferreiro, and Executive Committee member, Dr Anna Ambrosini. As they stepped away from their roles, we took the opportunity to ask them about their experiences and perspectives on the ENMC.

4.1 Interview with outgoing Research Director - Dr Ana Ferreiro

DR ANA FERREIRO is a neuromuscular neurologist at the Reference Center for Neuromuscular Diseases, Neuromyology Department, and is a translational scientist and Inserm Research Director at the Myology Research Center (Institute of Myology), both in the Pitié-Salpêtrière University Hospital in Paris. For over 25 years, she has combined clinical care with basic, translational, and therapeutic research on muscle diseases, with a particularly focus on congenital myopathies and desmin-related myopathies. Dr Ferreiro serves currently as an Executive Associate Editor for Neuromuscular Disorders and was the Research Director of the ENMC for 6 years (2019-2024).

How did you get involved with ENMC?

At the very beginning of my research career, in 1999, thanks to my mentor Prof Michel Fardeau. I was in Paris for a European PhD, searching to define phenotypically and genetically the congenital myopathy multi-minicore disease, which at the time involved recruiting a critical number of informative families. I still can hear him saying: 'There's a new initiative in the Netherlands which could help with this, we should apply'. So, we did, and I had the chance and the privilege to co-organise with Michel my first ENMC workshop in May 2000. The ENMC has been a constant reference for me since then, and has made a major difference not only for my research but in promoting progress in the field of neuromuscular diseases at the European and international levels.

What has been your experience of being a ENMC workshop organiser?

My first one was a really impactful experience, and one of the reasons why I have never left the neuromuscular field. I remember sitting for the first time around the table with a group of the best European experts, realising how unique and stimulating the exchanges and the cooperative spirit were and feeling that I had found my scientific family. I have lost count of the workshops which I have co-organised since then, and it has been a great experience every time. I think that the unique atmosphere created by the ENMC and the participants determination to work together for a common and greater good are really important to promote mutual trust and open exchanges, and to foster life-long collaborations.

What has your journey been like as the ENMC Research Director for the last six years?

It has been an intense and really rewarding journey, one of the best experiences of my professional life. I am very grateful for the opportunity to give back, by serving as Research Director, a bit of what the ENMC has given, to me and to the field, over the years. It was a joy working with the fantastic ENMC Research and Executive committees and with the ENMC office teams, in particular with Dr Alexandra Breukel who, as a Managing Director, was pivotal for transforming my ideas into realities. I also learned a lot about what is happening in neuromuscular diseases over the world, acquired a better knowledge of the different stakeholders in the landscape and had the privilege of working with representatives of many international Patient Advocacy Organisations, an extremely important driving force. Altogether, an exciting, enjoyable and stimulating experience, with lots of traveling and opportunities to exercise some diplomatic skills!

What has been the most important developments within the ENMC in the years you were the ENMC Research Director?

Actually, quite a lot has changed, hopefully for good. Going through the Coronavirus pandemic, with the impossibility to hold face-to-face workshops, required adapting our mode of functioning (thanks to the office and particularly Dr Breukel), introducing virtual meetings and more pre-workshop preparations. The latter have proven so effective and impactful that they have now become part of our standard requirements. An initiative which was in my mind for years and has become a reality thanks to the ENMC is the Mid-Career mentoring programme, which aims to support the emergence of the future leaders in the field. We expect that this will help them to open up more opportunities for those who come behind and thus attract the new generations, for whom we reinforced the Early Career programme. The Mentoring programme committee brought members of the Research and of the Executive committees of the ENMC to work together and get to know each other better, something I am very happy about. I also conceived the Themed workshops as a way to anticipate proactively and respond to the emerging needs in the field. This has now become, under the initiative of the Executive Committee, the axis of the General Assembly Meeting, a unique opportunity to bring together all the ENMC stakeholders. Finally, I think that the links between the ENMC and other international initiatives in the field, such as the European Academy of Neurology, TREAT-NMD, and the World Muscle Society, are now stronger than they were six years ago - a very positive development.

Why do you feel/think that the ENMC is still relevant/valuable in the current NMD field?

I think that it is more relevant than ever now that we are at a turning point in the field. The development of innovative therapies brings in new global challenges, risks and emerging needs which require a global, consensus answer. Also, attracting young people to the field is key for anticipating and building the future, and interacting with inspiring, leading NMD specialists in the ENMC context is a very effective driver in this sense. More generally, the ENMC climate of dialogue, cooperation and mutual support and trust, so important for scientific progress, is particularly precious and needed in the challenging and division-provoking times which we are living.

What ambitions do you see for the ENMC to achieve for the near future?

I have no doubt that with Ros Quinlivan as Research Director the ENMC will continue to be a driving force to facilitate the development of new therapies and the response to the new challenges that this entails (in terms of health risks, economics and policy considerations…). Promoting diversity and supporting and inspiring the next generations will be important. Finally, I trust that the ENMC will keep on working to bring multiple stakeholders together, including academia and patient advocacy organisations but also other important actors including industry, regulatory agencies or policy makers.
I will definitely enjoy to see that happening!

4.2 Interview with former Executive Committee member - Dr Anna Ambrosini

ANNA AMBROSINI has a PhD in pharmacology and was an academic researcher in the field of neurodegeneration until 2000. Since 2001 she has been working at Fondazione Telethon, the main Italian funding agency for rare genetic diseases, where she is currently Head of Research. Since 2018, she has also been Head of Research at Fondazione AriSLA, the Italian agency supporting amyotrophic lateral sclerosis research, of which Fondazione Telethon is a founding partner. She joined the ENMC Executive Committee in 2004 and contributed to its activities until December 2024.

How did you and/or Fondazione Telethon get involved with ENMC?

Fondazione Telethon was born in 1990 from the will of the Italian Muscular Dystrophy Association UILDM to support research on muscular dystrophies. As part of this activity, UILDM, which was one of the founders of ENMC back in 1990, asked Fondazione Telethon to take the lead on its behalf and represent both Italian entities in the ENMC governance. Fondazione Telethon has been part of the ENMC since 1992, when the ENMC legal entity was created.

What has been your experience of being a member of the ENMC Executive Committee for the last 20 years?

I have always enjoyed being part of the Executive Committee and having the opportunity to contribute to improving the ENMC's offer to the neuromuscular community. Listening to the voice of people living with a neuromuscular disease and fostering interactions within the stakeholder network has contributed greatly to my professional and personal growth, giving me a sense of belonging to a vibrant neuromuscular community that is strongly focused on the needs of patients.

What is the role of ENMC in the NMD field? And did the role change in the last 20 years?

The ENMC provides an independent hub for people involved in neuromuscular research to meet and discuss how to advance clinical research in the field, particularly addressing the most controversial issues related to neglected diseases and unmet needs. In its role as a facilitator of links between experts, the ENMC has been at the forefront in many areas, anticipating the needs and changes that have accompanied developments in the neuromuscular field over the last 30 years.

What has been the most important developments within the ENMC over the years (strategically and/or operationally)?

I can identify three main strategic phases. In the 1990s, the ENMC facilitated the creation of networks of clinical experts to promote understanding of the many neuromuscular diseases and to disseminate knowledge on clinical and genetic diagnostic criteria. After 2000, the creation of a support team of experts in biostatistics and clinical trial development set the stage for the development of the TREAT-NMD network, which involved the ENMC network of clinical scientists to accelerate trial readiness, starting from Duchenne muscular dystrophy and spinal muscular atrophy. Over the last 10-15 years,

the ENMC has further strengthened the relationship between the different stakeholders, ensuring that the voice of people living with a neuromuscular disease is heard in its workshops and becomes part of the new knowledge generated by the discussion.

Why do you feel/think it's so important to have a patient/patient representatives in a workshop?

Particularly in the past, clinicians often 'appeared' to listen to patients, but without actually being active listeners. The ENMC has fought against this old patronising attitude by discussing in depth with patient organisation representatives the level of involvement in research of people with a neuromuscular disease. This activity was also carried out through dedicated meetings and led to the development of the ENMC White Paper 2020 and the structuring of the active involvement of patients in the ENMC workshops. People who experience the disease in their daily lives bring their perspective on unmet medical needs and burden, often have different priorities from clinicians, and provide valuable information on how to measure the impact of treatments on their physiological function and quality of life.

What ambitions do you see for the ENMC to achieve for the near future?

I am particularly concerned about those diseases for which there is still no active clinical pipeline of experimental therapies. Facilitating the scientific discussion on these diseases and how to implement the lessons learned from the most studied ones, training a new generation of young clinical investigators, and monitoring the landscape to synergise with other ongoing networking initiatives are all aspects that the ENMC is engaged in, and I would like to see it continue to be a key player in this field.

5 Creating global awareness about ENMC workshops

5.1 Publication and dissemination of workshop outcomes

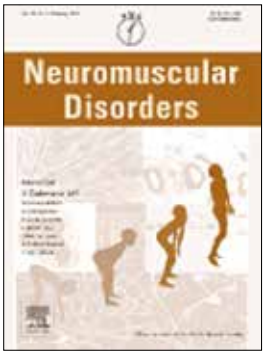
Informing patients and their families about the achievements of ENMC workshops is a key priority of the ENMC. For this purpose, a workshop lay report in English is written by workshop participants and published on the ENMC website within two weeks after the workshop. Lay reports are then translated into many different languages to increase their accessibility for people worldwide. The European partner organisations and other co-sponsors help to disseminate these translated lay reports via their local patient networks and the ENMC creates awareness on social media (LinkedIn: ENMC Non-profit group) during the workshops and whenever lay reports are published on its website.

The ENMC maintains an online archive of all workshops organised since 2000, which provides access for the general public to the outcome of the workshops over the years:

https://www.enmc.org/publications/workshop-reports/

Research community

Researchers, clinicians and healthcare providers who are active in the research field of rare neuromuscular disorders need to be able to read about the scientific results of ENMC workshops. Therefore, it is mandatory that workshop organisers submit a full workshop report to the journal Neuromuscular Disorders within 6 months of the workshop. Since 2021, Early-Career Researchers who made a significant contributions to the organisation of the workshop and the writing of the lay and full report, could become co-authors on the full report.



ENMC-workshop derived publications are cited 24% more than average (100%). In bibliometric terms this means that they have a "high impact" in the NMD research field.

5.2 International conferences in 2024

ENMC ambassadors have attended the following international congresses:

- 4th Scientific International Congress on SMA, Ghent, Belgium (14-16 March 2024).
- 8th International Congress Myology, Paris, France (22-25 April 2024).
- Peripheral Nerve Society Annual Meeting (PNS), Montreal, Canada (22-24 June 2024).
- European Academy of Neurology (EAN), Helsinki, Finland (29 June-2 July 2024).
- World Muscle Society Congress, Prague, Czech Republic (8-12 October 2024).
- ICNMD 2024, Perth, Australia (25-29 October 2024).

6 The ENMC Mid-Career Mentoring Programme

This programme has been developed for people who seek mentoring in order to acquire skills on their way toward becoming independent researchers and/or potential future leaders in the NMD field. These individuals typically are in the stage in their careers where they are developing their own research plans and have a proven track record in the neuromuscular field. They have established research teams and collaborative networks. The guidelines and the mentee and mentor forms can be found on the ENMC website:

Mid-Career Mentoring Programme

https://www.enmc.org/mid-career-mentoring-programme/introduction/

Deadline to apply is **July 1st** each year.

Drs Rossella Avagliano Trezza (Netherlands), Paloma Gonzalez-Perez (USA), Aurea Martins-Bach (UK) and Nicole Voet (Netherlands) were all selected as mentees as part of the inaugural Mentoring Programme (MP) in 2021. As they came to the conclusion of the MP, we sent them a survey and each mentee had an online meeting with Dr Ana Ferreiro and Prof. Ros Quinlivan (current and future Research Directors) to evaluate the programme. The survey showed that all mentees were very happy with this opportunity that had been provided by the ENMC and they would advocate the programme to others. They see improvement in the skills they wanted to develop or enhance through help of this programme. There were no signs of mismatch with the mentors nor with the communications with the ENMC. The mentees rated their experience highly. They all appreciated the online meeting, to elaborate further on their feedback and suggested some enhancements to the MP that we will be taking forward in the future. All mentees wanted to keep in touch with their mentors outside the MP.

Mentee 2024

DR LINDSAY ALFANO is a researcher specialising in the care and evaluation of patients with neuromuscular disorders. She is based at The Abigail Wexner Research Institute at Nationwide Children's Hospital, and is a principal investigator in the Center for Biobehavioral Health and Assistant Professor of Pediatrics at The Ohio State University.

Lindsay's mid-career mentors are Prof. A. Beggs, Prof. U. Schara-Schmidt and Prof. K. Wagner

7 Resources and financial management in 2024

Financial summary 2024

Annual accounts for the year 2024 were compiled in accordance with Guideline C1 for the reporting of small-sized non-profit organisations as published by the Dutch Accounting Standards Board. The financial accounts are drawn up in Euros.

In the summary table below, the overall income and expenditure over the year 2024 are shown in comparison with the figures for the previous financial year 2023.

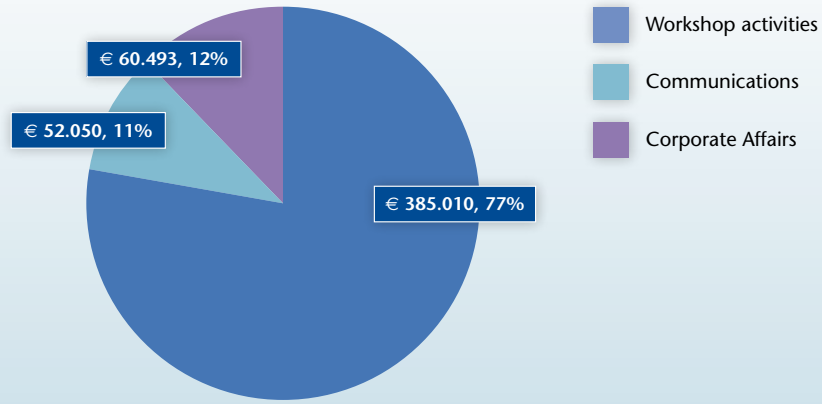
Statement of income and expenses for the year 2024 in Euros (€)		
	2024	2023
INCOME		
Full Partner contributions	231.000	231.000
Associated Partner contributions	25.000	25.000
Company Forum contributions	146.556	144.532
Other contributions	64.990	24.287
Total income	**467.546**	**424.819**
EXPENSES		
Personnel expenses	211.202	304.884
Rental expenses	11.893	11.701
Activity (workshop) expenses	223.367	89.980
Organisational expenses	51.091	71.603
Total operating expenses	**497.553**	**478.168**
Operating result	**– 30.007**	**– 53.349**
Interest income	8.148	2.713
Net result	**– 21.859**	**– 50.636**
APPROPRIATION OF RESULTS		
Transitional reserve	–	– 55.722
Transitional reserve - release	– 114.278	–
Global Travel Fund - allocation	114.278	–
Global Travel Fund - used	– 30.000	–
Other free reserves	8.141	5.086
CASH AT BANKS ON 31 DECEMBER	**637.208**	**607.414**

The distribution of income from the different ENMC supporters and the distribution of costs over the key accounts: workshop activities, corporate affairs and communications, are provided in the two diagrams.

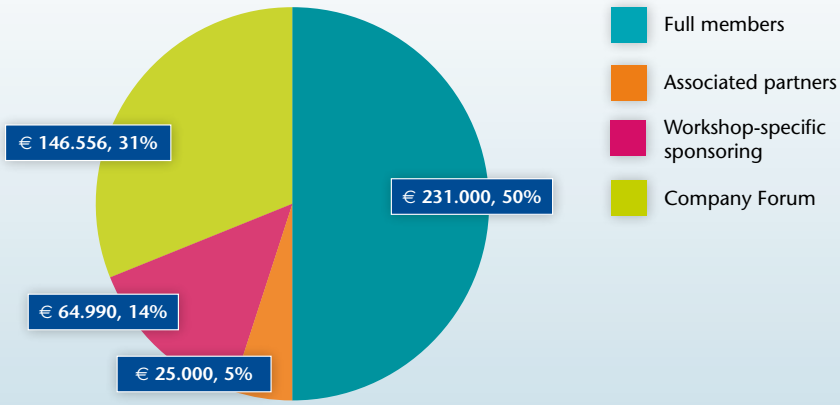
Opinion of the auditors

The independent accountants have verified and approved the annual accounts. For a full PDF version of the report of the annual accounts for 2024, please visit the ENMC website: **https://www.enmc.org/about-us/annual-report/**

Distribution of costs **2024**



Distribution of income **2024**



8 Governance in 2024

The European Neuromuscular Centre was founded as a non-profit organisation on 24 November 1992 under Dutch law. The foundation is supported by financial contributions of European patient organisations for neuromuscular disorders and many other related organisations. The statutory location is in Baarn, The Netherlands, in the building of Spierziekten Nederland.

8.1 The ENMC Executive Committee

The ENMC is governed by an Executive Committee consisting of representatives of ENMC partner organisations.

Composition of the ENMC Executive Committee on 31 December 2024
Dr K. Adcock (vice-Chair, United Kingdom)
Dr D. Baldessari (Italy)
Dr S. van den Berge (The Netherlands)
Mr H. Ib Jørgensen (Denmark)
Dr I. Meijer (Chair, The Netherlands)
Dr A. Méjat (France)
Prof. U. Schara-Schmidt (Germany)
Dr R. Willmann (Switzerland)

8.2 The ENMC Research Committee

The ENMC Research Committee is responsible for reviewing the scientific content and quality of the workshop applications and advises the Executive Committee on awarding the grants for ENMC workshops.

Composition of the ENMC Research Committee on 31 December 2024
Dr A. Buj-Bello (France)
Dr A. Ferreiro (Chair, France)
Prof. Dr N. Goemans (Belgium)
Prof. Dr E. Gomes (Portugal)
Prof. Dr C. Kornblum (Germany)
Prof. C. Ottenheijm (The Netherlands)
Dr C. Paradas (Spain)
Dr D. Pareyson (Italy)
Prof. R. Quinlivan (United Kingdom)
Prof. G. Tasca (United Kingdom)
Prof. M. Weber (Switzerland)

ENMC Executive Committee meeting in November 2024 with the ENMC Research Director (RD) Dr Ana Ferreiro and future RD Prof. Ros Quinlivan.

ENMC Research Committee meeting in November 2024.

8.3 The ENMC Office

The office takes care of the daily business of the ENMC.

ENMC Office staff on 31 December 2024
Ms P. van Dongen (Programme Manager)
Ms W. Hinloopen (Operational Manager)
Ms T. van Esch (Freelance Workshop Assistant)

ENMC office staff, Wilma Hinloopen and Patricia van Dongen

8.4 Transfer of the Fondazione Telethon ETS representative

After representing the Fondazione Telethon ETS at the ENMC Executive Committee for 20 years (from 2004-2024), Dr Anna Ambrosini passed her task to Dr Danila Baldessari in November 2024.

Anna has played a vital role in the governance of the ENMC, bringing a sharp, analytical perspective and asking thought-provoking questions. She demonstrated strong leadership in designing, preparing, and organising the meeting on "The Position of the Neuromuscular Patient in Shared Decision-Making," held in Milan in January 2018. The vision established during that meeting remains relevant today. Her perseverance was also key in finalising several publications as well as in drafting the White Paper*, which continues to serve as an excellent guide on patient engagement. It emphasises how patient involvement has long been recognised as a crucial issue and highlights the importance of empowering patients and patient organisations to define how and when they wish to participate in research and healthcare, shaping the co-creation process for the future.

Anna has always stayed true to the core mission of ENMC. While exploring new opportunities and directions, she ensured that ENMC remained focussed on its primary goal - organising high-quality, impactful workshops renowned in the neuromuscular field.

We extend our deepest gratitude to Anna for her invaluable contributions, unwavering dedication, and lasting commitment to ENMC over the years.

A "Thank You" gift from Dr Ingeborg Meijer on behalf of ENMC to Dr Anna Ambrosini during the EC meeting in November 2024.

** https://www.enmc.org/patient-participation/enmc-publications-about-patient-involvement-in-the-neuromuscular-field/*

Dr Danila Baldessari is Research Programme Manager at the Scientific Office of Fondazione Telethon. Currently, Danila is dedicated to neuromuscular research area, where she coordinates the call for neuromuscular clinical research projects and programmes, oversees research monitoring and supports activities for clinical research networks, project development and the Italian registries supported by the Fondazione Telethon Registry Association. Danila has also focused on research impact and strategic analysis, as well as the implementation of Open Access and Research Integrity policies for Fondazione Telethon investigators.
The ENMC welcomes her to the Executive Committee and looks forward to a great collaboration.

Dr Danila Baldessari

8.5 Goodbye from Dr Ana Ferreiro as ENMC Research Director

The term of Dr Ana Ferreiro ended on 31th December 2024 after 6 years of fulfilling the role of ENMC Research Director. The ENMC greatly thanks her for taking on the ambassador role, as she was able to connect ENMC with new clinicians, researchers, and organisations. We also appreciate her excellent chairmanship of the Research Committee, as well as her support and advice in implementing innovations like the ENMC Mid-Career Mentoring Programme.

One of the presents that Dr Ana Ferreiro received was a booklet with photos of all her ENMC activities as a workshop participant, organiser and Research Director in the last 20 years.

9 A special thank-you to all our partners and supporters

It is thanks to the continuous support of the eight European patient organisations that the ENMC can facilitate and organise, on average, eight workshops per year. With support from additional partner organisations, such as condition-specific associations, associated partners and members of the ENMC Company Forum, we are also able to invite participants from non-European countries and facilitate the attendance of Early-Career Researchers, patients and patient representatives.

ENMC full partners

AFMTÉLÉTHON
INNOVER POUR GUERIR

Deutsche Gesellschaft für
Muskelkranke e.V. DGM

FONDAZIONE
Telethon

fsrmm
fondation suisse de recherche sur les maladies musculaires
fondazione svizzera per la ricerca sulle malattie muscolari
schweiz. stiftung für die erforschung der muskelkrankheiten

MUSCULAR DYSTROPHY UK
OUR MUSCLES MATTER

MUSKELSVINDFONDEN

Prinses Beatrix
Spierfonds

SPIERZIEKTEN NEDERLAND

ENMC associated partners

Finnish Neuromuscular Disorders Association

Österreichische MuskelForschung

SMA Europe

WMS World Muscle Society

World Duchenne Organization UPPMD

Members of the Company Forum

Amicus Therapeutics

astellas Gene Therapies

Biogen

IONIS

Lupin Neurosciences

Novartis

revvity

Roche

sanofi

santhera

Workshop-specific sponsors in 2024

10 Looking forward to 2025 and beyond

10.1 Workshops in 2025

Five ENMC workshops are scheduled to take place in Q1 of 2025 and at least three further workshops are planned later in the year. (see table below).

Two review rounds for workshop applications are scheduled in 2025: one in the spring (submission deadline 1 March 2025) and one in the autumn (submission deadline 1 October 2025). The workshops that are selected at these review rounds will all be planned in 2025 or 2026.

For updates please visit the ENMC website: **https://www.enmc.org/workshops/upcoming-workshops/**

Preliminary ENMC programme 2024

Workshop no. and date	Topic	Workshop leaders
Workshop no. 283 17-19 January 2025	Establishing expert care recommendations for LAMA2-RDs: a prototype for the development of congenital muscular dystrophy subtype-specific care guidelines	Dr R. Foley, Prof. A. Klein, Dr A. Sarkozy, Dr A. Zambon
Workshop no. 284 24-26 January 2025	Cognitive and behavioral abnormalities in pediatric DM1 – what should we measure in preparation for clinical trials?	Prof. V. Sansone, Prof. N. Johnson
Workshop no. 285 31 January - 2 February 2025	SMN-related neurodevelopmental disorder: type 1 Spinal Muscular Atrophy and the brain	Prof. Quijano-Roy, Prof. G. Baranello, Dr D. Gómez Andrés, Prof. M. Farrar
Workshop no. 286 7-9 March 2025	Muscle imaging: Artificial Intelligence, Automatic Segmentation and Imaging Data Sharing in Neuromuscular Disease	Prof. V. Straub, Dr H. Kan, Dr J. Warman Chardon, Prof. J. Vissing
Workshop no. 287 28-30 March 2025	Harmonisation and federated analysis of Myotonic Dystrophy registries to model heterogeneous disease trajectories	Prof. P. 't Hoen, Prof. C. Faber, Prof. N. Johnson, Dr G. Bassez
Workshop no. 288 16-18 May 2025	Towards better diagnosing, understanding and treating gastrointestinal symptoms in myotonic dystrophy	Dr H. Braakman, Prof. L. Pastorelli, Prof. G. Meola, Prof. B. Schoser
Workshop no. 289 26-28 September 2025	Assessing and managing emerging AAV related toxicities after gene therapy for neuromuscular disorders	Prof. F. Muntoni, Prof. H. Büning, Prof. C. Bönnemann
Workshop no. 290 21-23 November 2025	Developing Standards of Care for patients with Becker Muscular Dystrophy (BMD)	Prof. M. Guglieri, Prof. E. Pegoraro, Dr E. Niks, Prof. R. Quinlivan

10.2 International conferences in 2025

ENMC ambassadors will attend the following international congresses.

- Congress of the Medical Scientific Advisory Board of the German Muscular Dystrophy Society DGM e.V., Giessen, Germany (19-21 March 2025).
- 18th UK Neuromuscular Translational Research Conference 2025, Newcastle upon Tyne, United Kingdom (15-16 April 2025).
- Peripheral Nerve Society Annual Meeting (PNS), Edinburgh, United Kingdom (17-20 May 2025).
- European Academy of Neurology (EAN), Helsinki, Finland (21-24 June 2025).
- 16th European Paediatric Neurology Society Congress, Munich, Germany (8-12 July 2025).
- World Muscle Society Congress, Vienna, Austria (7-11 October 2025).

10.3 Welcome to our new Research Director

Prof. Ros Quinlivan will become the new ENMC Research Director as of 1st January 2025.
We congratulate Ros on her new task within the organisation and looking forward to working with her. She is Professor of Neuromuscular Disease at the National Hospital for Neurology and Neurosurgery, UCL Institute of Neurology, Queen Square, London, where she leads on transition and managing young adults with neuromuscular disease.

We will be working on themes such as encouragement of early career researchers to join the ENMC network and patient participation in ENMC activities. Ros will accompany the ENMC office at some NMD meetings and at ENMC workshops to meet you in person.

Prof. Ros Quinlivan

10.4 Budget for 2025

This table presents the budget forecast for 2025 as of 31 December 2024.

Budget 2025 in Euros (€)	Actuals 2024	Budget 2025
INCOME		
Full Partner contributions	231.000	242.550
Associated Partner contributions	25.000	25.000
Company Forum contributions	146.556	120.000
Other contributions	64.990	45.000
Total income	**467.546**	**432.550**
EXPENSES		
Personnel expenses	211.202	214.000
Rental expenses	11.983	12.000
Activity expenses	223.367	225.000
Organisational expenses	51.091	68.000
Total operating expenses	**497.553**	**519.000**
Operational result	– **30.007**	– 86.450
Interest income	8.148	6.500
NET RESULT	– **21.859**	– **79.950**

The ENMC was present at the World Muscle Society congress with a booth. From left to right: Wilma Hinloopen, Prof. Ulrike Schara-Schmidt (EC member), Dr Simone van den Berge (EC member), Dr Ana Ferreiro (Research Director), and Patricia van Dongen.

Colophon

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Wilma Hinloopen
Ingeborg Meijer

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